



# HUMAN GENOME STUDIES

## *science for the 21st Century*

*will involve the continued research into human genome*

Los Alamos has been well known for its expertise in human genetics for decades. In 1982, the Laboratory established GenBank, a database that serves as a national repository for genetic sequence information. The following year, Los Alamos helped lay the foundation for the Human Genome Project when it created the first DNA libraries from flow-sorted chromosomes, which comprise the human DNA.

The Human Genome Project is a joint effort by the Department of Energy and the National Institutes of Health to identify all of the approximately 140,000 genes in human DNA and determine the sequences (order) of the three billion chemical base pairs that make up the human genome.

As part of their work in the Human Genome Project, Los Alamos researchers discovered the first detailed information on the structure and importance of telomeres (the ends of chromosomes) and provided new information on the key roles in cell division played by the specialized chromosomal regions known as centromeres. In 1995, Los Alamos produced the first high-resolution physical map of chromosome 16, outlining the order of cloned DNA fragments that together reveal the sequence of the entire chromosome.

In 1996, Los Alamos joined forces with other DOE national laboratories to combine their genome centers into a single virtual organization called the Joint Genome Institute. Los Alamos has the lead role in ensuring sequencing data quality for the institute. In November 1999, the JGI participants celebrated with other large sequencing centers worldwide the completion of sequencing one billion of the human genome's three billion base pairs.

The JGI is part of a consortium of five international genome institutes working together to sequence the entire genome by 2003. The task could be completed sooner, as Los Alamos and others are continually developing more efficient sequencing technologies. The work of the JGI is responsible for sequencing chromosomes that contain genes involved in diabetes, atherosclerosis, asthma, leukemia, certain cancers, schizophrenia and other disorders.

Today Los Alamos performs major research in functional genomics — the study of the function of genes and their interactions with each other — and structural genomics — the study of the physical organization of the proteins encoded by families of genes and the relationships between structure and function of those proteins.

The information derived from genetic research at Los Alamos helps physicians to better understand human biology and health care. Knowledge about the effects of DNA variations among individuals may help researchers better diagnose, treat and possibly even eradicate the more than 4,000 genetically inherited human disorders. The genetics work at Los Alamos might someday even help solve problems in such diverse fields as energy, agriculture and environmental cleanup.

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